



ETFB gene

electron transfer flavoprotein beta subunit

Normal Function

The *ETFB* gene provides instructions for making one part (the beta subunit) of an enzyme called electron transfer flavoprotein. This enzyme is normally active in the mitochondria, the energy-producing centers in cells. Electron transfer flavoprotein is involved in the process by which fats and proteins are broken down to produce energy.

Health Conditions Related to Genetic Changes

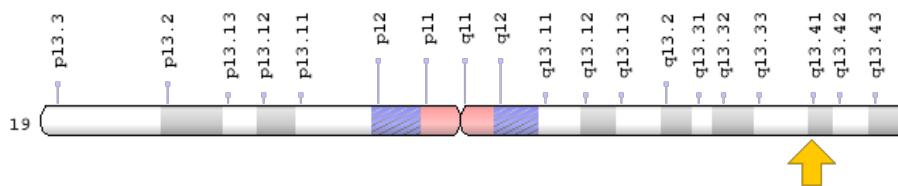
glutaric acidemia type II

Some mutations in the *ETFB* gene prevent the production of the electron transfer flavoprotein enzyme. Other mutations result in the production of a defective enzyme that cannot fulfill its role in the series of reactions (metabolic pathways) that break down fats and proteins. This enzyme deficiency allows these nutrients, as well as compounds created as the nutrients are partially broken down, to build up to abnormal levels, especially when the body is under stress. Toxic products of incomplete metabolism damage cells in many body systems, resulting in the signs and symptoms of glutaric acidemia type II.

Chromosomal Location

Cytogenetic Location: 19q13.41, which is the long (q) arm of chromosome 19 at position 13.41

Molecular Location: base pairs 51,345,155 to 51,366,418 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- electron transfer flavoprotein, beta polypeptide
- electron-transfer-flavoprotein, beta polypeptide
- ETFB_HUMAN
- FP585

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ETFB%5BTIAB%5D%29+OR+%28MADD%5BTIAB%5D%29+OR+%28electron+transfer+flavoprotein+beta+subunit%5BTIAB%5D%29+OR+%28electron+transfer+flavoprotein+beta-subunit%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- ELECTRON TRANSFER FLAVOPROTEIN, BETA POLYPEPTIDE
<http://omim.org/entry/130410>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ETFB%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3482
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/2109>
- UniProt
<http://www.uniprot.org/uniprot/P38117>

Sources for This Summary

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